#4/B

~~[illegible]~~

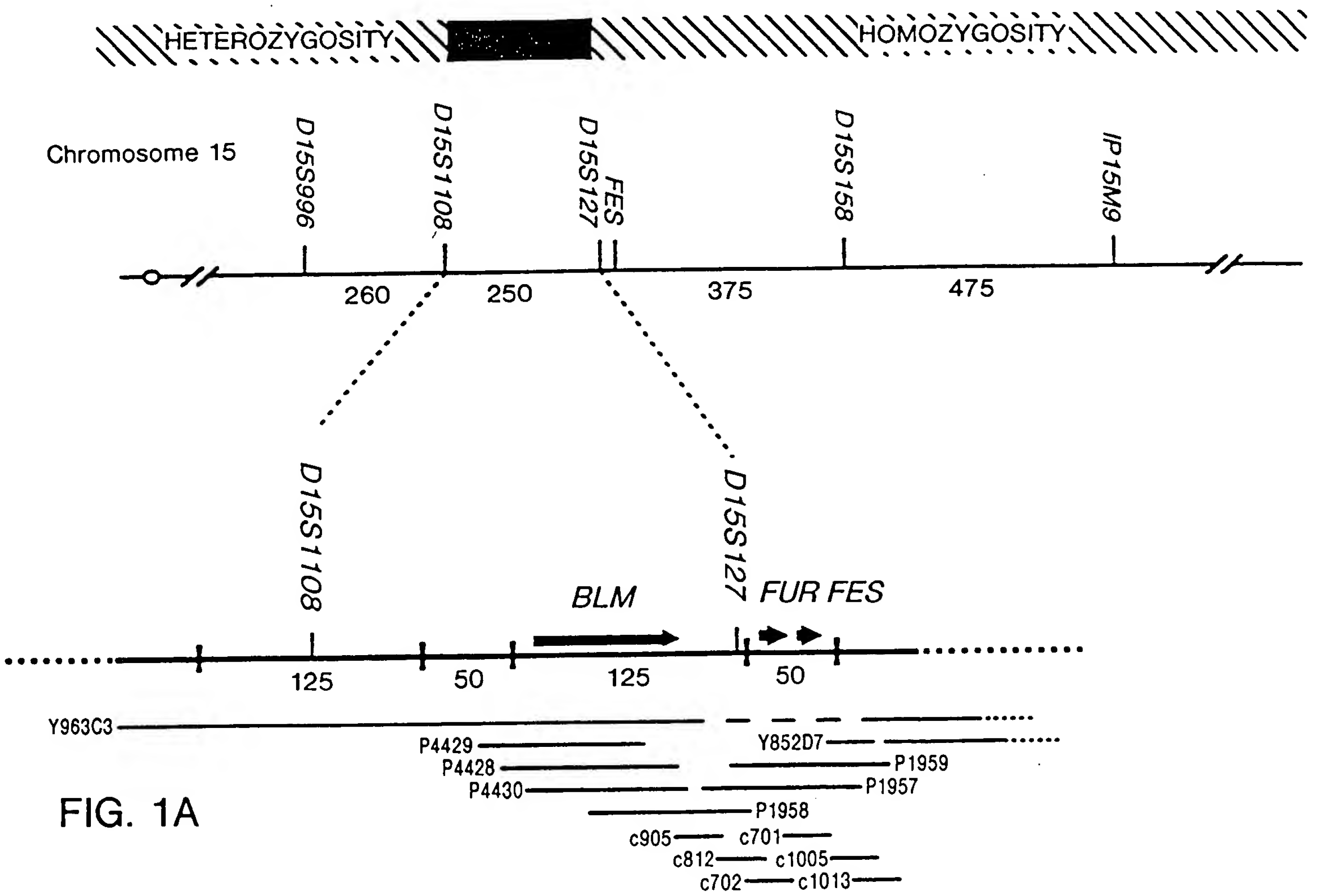
HETEROZYGOSITY
HOMOZYGOSITY
Chromosome 15
D15S996
D15S1108
D15S127
FES
D15S158
IP15M9
260
250
375
475
D15S1108
D15S127
BLM
FUR FES
125
50
125
50
Y963C3
Y852D7
P4429
P4428
P4430
P1959
P1957
P1958
c905
c701
c812
c1005
c702
c1013

FIG. 1A

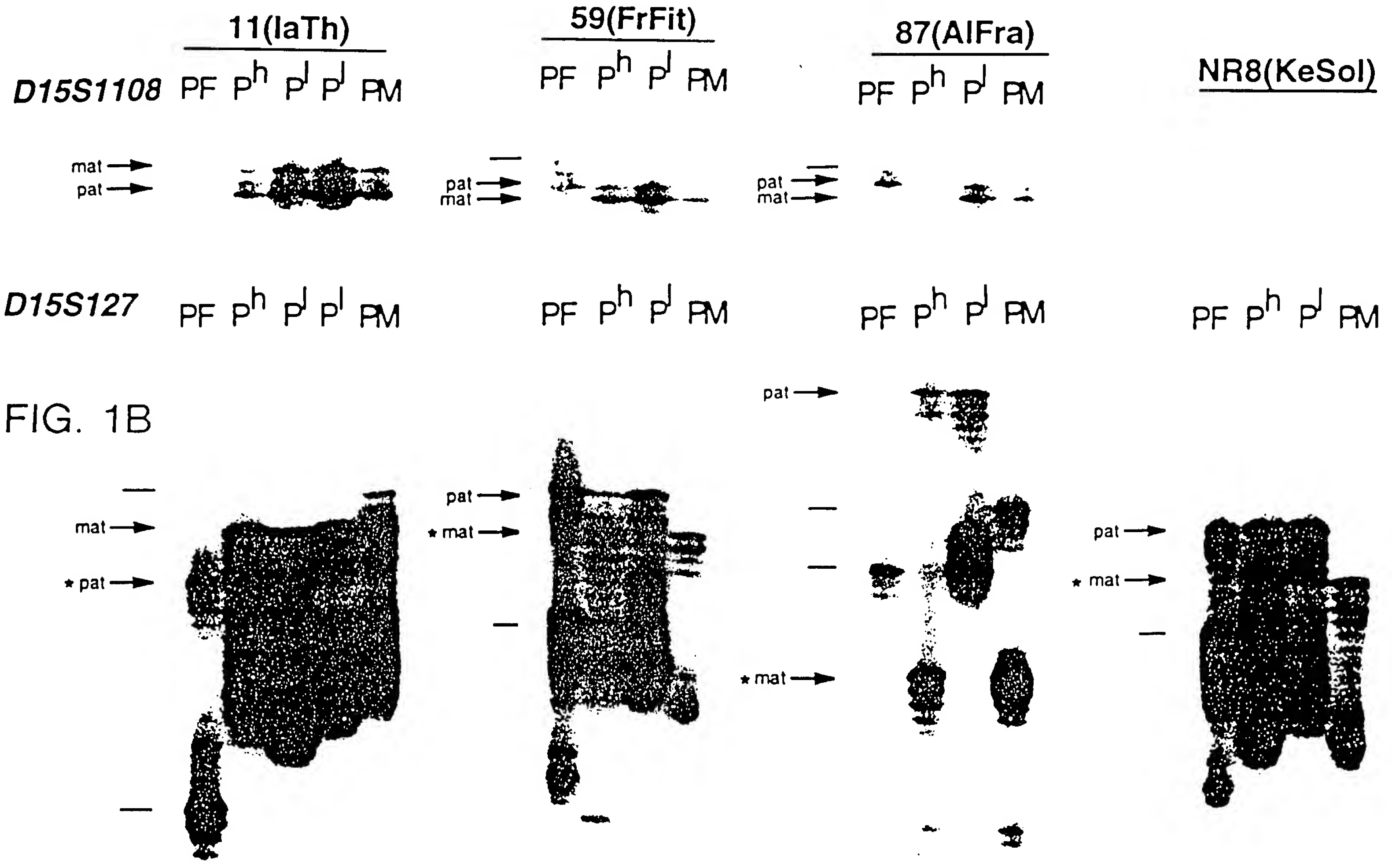
11(IaTh)
59(FrFit)
87(AlFra)
NR8(KeSol)
D15S1108
PF Ph PJ PJ PM
PF Ph PJ PM
PF Ph PJ PM
mat
pat
pat
mat
pat
mat
D15S127
PF Ph PJ PJ PM
PF Ph PJ PM
PF Ph PJ PM
PF Ph PJ PM
mat
* pat
pat
* mat
pat
* mat
pat
* mat

FIG. 1B

```
    gcgcggcggccgtggttgcggcgcgggaagtttggatcctggttccgtccgctaggagtctgcgtgcgaggattATGGCT   80
                                                                               M  A
    GCTGTTCCTCAAAATAATCTACAGGAGCAACTAGAACGTCACTCAGCCAGAACACTTAATAATAAATTAAGTCTTTCAAA  160
  3 A  V  P  Q  N  N  L  Q  E  Q  L  E  R  H  S  A  R  T  L  N  N  K  L  S  L  S  K
    ACCAAAATTTTCAGGTTTCACTTTTAAAAAGAAAACATCTTCAGATAACAATGTATCTGTAACTAATGTGTCAGTAGCAA  240
 30  P  K  F  S  G  F  T  F  K  K  K  T  S  S  D  N  N  V  S  V  T  N  V  S  V  A
    AAACACCTGTATTAAGAAATAAAGATGTTAATGTTACCGAAGACTTTTCCTTCAGTGAACCTCTACCCAACACCACAAAT  320
 56 K  T  P  V  L  R  N  K  D  V  N  V  T  E  D  F  S  F  S  E  P  L  P  N  T  T  N
    CAGCAAAGGGTCAAGGACTTCTTTAAAAATGCTCCAGCAGGACAGGAAACACAGAGAGGTGGATCAAAATCATTATTGCC  400
 83  Q  Q  R  V  K  D  F  F  K  N  A  P  A  G  Q  E  T  Q  R  G  G  S  K  S  L  L  P
    AGATTTCTTGCAGACTCCGAAGGAAGTTGTATGCACTACCCAAAACACACCAACTGTAAAGAAATCCCGGGATACTGCTC  480
110  D  F  L  Q  T  P  K  E  V  V  C  T  T  Q  N  T  P  T  V  K  K  S  R  D  T  A
    TCAAGAAATTAGAATTTAGTTCTTCACCAGATTCTTTAAGTACCATCAATGATTGGGATGATATGGATGACTTTGATACT  560
135 L  K  K  L  E  F  S  S  S  P  D  S  L  S  T  I  N  D  W  D  D  M  D  D  F  D  T
    TCTGAGACTTCAAAATCATTTGTTACACCACCCCAAAGTCACTTTGTAAGAGTAAGCACTGCTCAGAAATCAAAAAAGGG  640
163  S  E  T  S  K  S  F  V  T  P  P  Q  S  H  F  V  R  V  S  T  A  Q  K  S  K  K  G
    TAAGAGAAACTTTTTTAAAGCACAGCTTTATACAACAAACACAGTAAAGACTGATTTGCCTCCACCCTCCTCTGAAAGCG  720
190  K  R  N  F  F  K  A  Q  L  Y  T  T  N  T  V  K  T  D  L  P  P  P  S  S  E  S
    AGCAAATAGATTTGACTGAGGAACAGAAGGATGACTCAGAATGGTTAAGCAGCGATGTGATTTGCATCGATGATGGCCCC  800
216 E  Q  I  D  L  T  E  E  Q  K  D  D  S  E  W  L  S  S  D  V  I  C  I  D  D  G  P
    ATTGCTGAAGTGCATATAAATGAAGATGCTCAGGAAAGTGACTCTCTGAAAACTCATTTGGAAGATGAAAGAGATAATAG  880
243  I  A  E  V  H  I  N  E  D  A  Q  E  S  D  S  L  K  T  H  L  E  D  E  R  D  N  S
    CGAAAAGAAGAAGAATTTGGAAGAAGCTGAATTACATTCAACTGAGAAAGTTCCATGTATTGAATTTGATGATGATGATT  960
270  E  K  K  K  N  L  E  E  A  E  L  H  S  T  E  K  V  P  C  I  E  F  D  D  D  D
    ATGATACGGATTTTGTTCCACCTTCTCCAGAAGAAATTATTTCTGCTTCTTCTTCCTCTTCAAAATGCCTTAGTACGTTA 1040
296 Y  D  T  D  F  V  P  P  S  P  E  E  I  I  S  A  S  S  S  S  S  K  C  L  S  T  L
    AAGGACCTTGACACATCTGACAGAAAAGAGGATGTTCTTAGCACATCAAAAGATCTTTTGTCAAAACCTGAGAAAATGAG 1120
```

FIG. 2A

```
323  K  D  L  D  T  S  D  R  K  E  D  V  L  S  T  S  K  D  L  L  S  K  P  E  K  M  S
    TATGCAGGAGCTGAATCCAGAAACCAGCACAGACTGTGACGCTAGACAGATAAGTTTACAGCAGCAGCTTATTCATGTGA 1200
350   M  Q  E  L  N  P  E  T  S  T  D  C  D  A  R  Q  I  S  L  Q  Q  Q  L  I  H  V
    TGGAGCACATCTGTAAATTAATTGATACTATTCCTGATGATAAACTGAAACTTTTGGATTGTGGGAACGAACTGCTTCAG 1280
376 M  E  H  I  C  K  L  I  D  T  I  P  D  D  K  L  K  L  L  D  C  G  N  E  L  L  Q
    CAGCGGAACATAAGAAGGAAACTTCTAACGGAAGTAGATTTTAATAAAAGTGATGCCAGTCTTCTTGGCTCATTGTGGAG 1360
403  Q  R  N  I  R  R  K  L  L  T  E  V  D  F  N  K  S  D  A  S  L  L  G  S  L  W  R
    ATACAGGCCTGATTCACTTGATGGCCCTATGGAGGGTGATTCCTGCCCTACAGGGAATTCTATGAAGGAGTTAAATTTTT 1440
430   Y  R  P  D  S  L  D  G  P  M  E  G  D  S  C  P  T  G  N  S  M  K  E  L  N  F
    CACACCTTCCCTCAAATTCTGTTTCTCCTGGGACTGTTTACTGACTACCACCCTAGGAAAGACAGGATTCTCTGCCACC 1520
456 S  H  L  P  S  N  S  V  S  P  G  D  C  L  L  T  T  T  L  G  K  T  G  F  S  A  T
    AGGAAGAATCTTTTTGAAAGGCCTTTATTCAATACCCATTTACAGAAGTCCTTTGTAAGTAGCAACTGGGCTGAAACACC 1600
483  R  K  N  L  F  E  R  P  L  F  N  T  H  L  Q  K  S  F  V  S  S  N  W  A  E  T  P
              ◆
    AAGACTAGGAAAAAAAAAATGAAAGCTCTTATTTCCCAGGAAATGTTCTCACAAGCACTGCTGTGAAAGATCAGAATAAAC 1680
510   R  L  G  K  K  N  E  S  S  Y  F  P  G  N  V  L  T  S  T  A  V  K  D  Q  N  K
    ATACTGCTTCAATAAATGACTTAGAAAGAGAAACCCAACCTTCCTATGATATTGATAATTTTGACATAGATGACTTTGAT 1760
536 H  T  A  S  I  N  D  L  E  R  E  T  Q  P  S  Y  D  I  D  N  F  D  I  D  D  F  D
    GATGATGATGACTGGGAAGACATAATGCATAATTTAGCAGCCAGCAAATCTTCCACAGCTGCCTATCAACCCATCAAGGA 1840
563  D  D  D  D  W  E  D  I  M  H  N  L  A  A  S  K  S  S  T  A  A  Y  Q  P  I  K̇  E
    AGGTCGGCCAATTAAATCAGTATCAGAAAGACTTTCCTCAGCCAAGACAGACTGTCTTCCAGTGTCATCTACTGCTCAAA 1920
590   Ġ  Ṙ  Ṗ  I  K̇  Ṡ  V  S  E  Ṙ  L̇  S  S  A  K  T  D  C  L  P  V  S  Ṡ  T  Ȧ  Q
    ATATAAACTTCTCAGAGTCAATTCAGAATTATACTGACAAGTCAGCACAAAATTTAGCATCCAGAAATCTGAAACATGAG 2000
616 Ṅ  I  N  Ḟ  S  Ė  S  I  Q  N  Y  T  D  K  S  A  Q̇  N  L̇  A  S  Ṙ  N  L  K  H  E
    CGTTTCCAAAGTCTTAGTTTTCCTCATACAAAGGAAATGATGAAGATTTTTCATAAAAAATTTGGCCTGCATAATTTTAG 2080
643  R  F  Q̇  Ṡ  L̇  S  F  Ṗ  H  Ṫ  K  E  M  M  K  I  Ḟ  Ḣ  K  K  F  G  L  H  N  F  R
    AACTAATCAGCTAGAGGCGATCAATGCTGCACTGCTTGGTGAAGACTGTTTTATCCTGATGCCGACTGGAGGTGGTAAGA 2160
670   T  N  Q  L  E  A  I  N  A  A  L  L  G  E  D  C  F  I  L  M  P  T  G  G  G  K
    GTTTGTGTTACCAGCTCCCTGCCTGTGTTTCTCCTGGGGTCACTGTTGTCATTTCTCCCTTGAGATCACTTATCGTAGAT 2240
```

FIG. 2B

```
696 S  L  C  Y  Q  L  P  A  C  V  S  P  G  V  T  V  V  I  S  P  L  R  S  L  I  V  D
    CAAGTCCAAAAGCTGACTTCCTTGGATATTCCAGCTACATATCTGACAGGTGATAAGACTGACTCAGAAGCTACAAATAT 2320
723  Q  V  Q  K  L  T  S  L  D  I  P  A  T  Y  L  T  G  D  K  T  D  S  E  A  T  N  I
    TTACCTCCAGTTATCAAAAAAAGACCCAATCATAAAACTTCTATATGTCACTCCAGAAAAGATCTGTGCAAGTAACAGAC 2400
750  Y  L  Q  L  S  K  K  D  P  I  I  K  L  L  Y  V  T  P  E  K  I  C  A  S  N  R
    TCATTTCTACTCTGGAGAATCTCTATGAGAGGAAGCTCTTGGCACGTTTTGTTATTGATGAAGCACATTGTGTCAGTCAG 2480
776 L  I  S  T  L  E  N  L  Y  E  R  K  L  L  A  R  F  V  I  D  E  A  H  C  V  S  Q
    TGGGGACATGATTTTCGTCAAGATTACAAAAGAATGAATATGCTTCGCCAGAAGTTTCCTTCTGTTCCGGTGATGGCTCT 2560
803  W  G  H  D  F  R  Q  D  Y  K  R  M  N  M  L  R  Q  K  F  P  S  V  P  V  M  A  L
    TACGGCCACAGCTAATCCCAGGGTACAGAAGGACATCCTGACTCAGCTGAAGATTCTCAGACCTCAGGTGTTTAGCATGA 2640
830  T  A  T  A  N  P  R  V  Q  K  D  I  L  T  Q  L  K  I  L  R  P  Q  V  F  S  M
    GCTTTAACAGACATAATCTGAAATACTATGTATTACCGAAAAAGCCTAAAAAGGTGGCATTTGATTGCCTAGAATGGATC 2720
856 S  F  N  R  H  N  L  K  Y  Y  V  L  P  K  K  P  K  K  V  A  F  D  C  L  E  W  I
    AGAAAGCACCACCCATATGATTCAGGGATAATTTACTGCCTCTCCAGGCGAGAATGTGACACCATGGCTGACACGTTACA 2800
883  R  K  H  H  P  Y  D  S  G  I  I  Y  C  L  S  R  R  E  C  D  T  M  A  D  T  L  Q
    GAGAGATGGGCTCGCTGCTCTTGCTTACCATGCTGGCCTCAGTGATTCTGCCAGAGATGAAGTGCAGCAGAAGTGGATTA 2880
910  R  D  G  L  A  A  L  A  Y  H  A  G  L  S  D  S  A  R  D  E  V  Q  Q  K  W  I
    ATCAGGATGGCTGTCAGGTTATCTGTGCTACAATTGCATTTGGAATGGGGATTGACAAACCGGACGTGCGATTTGTGATT 2960
936 N  Q  D  G  C  Q  V  I  C  A  T  I  A  F  G  M  G  I  D  K  P  D  V  R  F  V  I
    CATGCATCTCTCCCTAAATCTGTGGAGGGTTACTACCAAGAATCTGGCAGAGCTGGAAGAGATGGGGAAATATCTCACTG 3040
963  H  A  S  L  P  K  S  V  E  G  Y  Y  Q  E  S  G  R  A  G  R  D  G  E  I  S  H  C
    CCTGCTTTTCTATACCTATCATGATGTGACCAGACTGAAAAGACTTATAATGATGGAAAAAGATGGAAACCATCATACAA 3120
990  L  L  F  Y  T  Y  H  D  V  T  R  L  K  R  L  I  M  M  E  K  D  G  N  H  H  T
    GAGAAACTCACTTCAATAATTTGTATAGCATGGTACATTACTGTGAAAATATAACGGAATGCAGGAGAATACAGCTTTTG 3200
1016 R  E  T  H  F  N  N  L  Y  S  M  V  H  Y  C  E  N  I  T  E  C  R  R  I  Q  L  L
    GCCTACTTTGGTGAAAATGGATTTAATCCTGATTTTTGTAAGAAACACCCAGATGTTTCTTGTGATAATTGCTGTAAAAC 3280
1043 A  Y  F  G  E  N  G  F  N  P  D  F  C  K  K  H  P  D  V  S  C  D  N  C  C  K  T
```

FIG. 2C

AAAGGATTATAAAACAAGAGATGTGACTGACGATGTGAAAAGTATTGTAAGATTTGTTCAAGAACATAGTTCATCACAAG 3360
1070 K D Y K T R D V T D D V K S I V R F V Q E H S S S Q
GAATGAGAAATATAAAACATGTAGGTCCTTCTGGAAGATTTACTATGAATATGCTGGTCGACATTTTCTTGGGGAGTAAG 3440
1096 G M R N I K H V G P S G R F T M N M L V D I F L G S K
AGTGCAAAAATCCAGTCAGGTATATTTGGAAAAGGATCTGCTTATTCACGACACAATGCCGAAAGACTTTTTAAAAAGCT 3520
1123 S A K I Q S G I F G K G S A Y S R H N A E R L F K K L
GATACTTGACAAGATTTTGGATGAAGACTTATATATCAATGCCAATGACCAGGCGATCGCTTATGTGATGCTCGGAAATA 3600
1150 I L D K I L D E D L Y I N A N D Q A I A Y V M L G N
AAGCCCAAACTGTACTAAATGGCAATTTAAAGGTAGACTTTATGGAAACAGAAAATTCCAGCAGTGTGAAAAAACAAAAA 3680
1176 K A Q T V L N G N L K V D F M E T E N S S S V K K Q K
GCGTTAGTAGCAAAAGTGTCTCAGAGGGAAGAGATGGTTAAAAAATGTCTTGGAGAACTTACAGAAGTCTGCAAATCTCT 3760
1203 A L V A K V S Q R E E M V K K C L G E L T E V C K S L
GGGGAAAGTTTTTGGTGTCCATTACTTCAATATTTTTAATACCGTCACTCTCAAGAAGCTTGCAGAATCTTTATCTTCTG 3840
1230 G K V F G V H Y F N I F N T V T L K K L A E S L S S
ATCCTGAGGTTTTGCTTCAAATTGATGGTGTTACTGAAGACAAACTGGAAAAATATGGTGCGGAAGTGATTTCAGTATTA 3920
1256 D P E V L L Q I D G V T E D K L E K Y G A E V I S V L
CAGAAATACTCTGAATGGACATCGCCAGCTGAAGACAGTTCCCCAGGGATAAGCCTGTCCAGCAGCAGAGGCCCCGGAAG 4000
1283 Q K Y S E W T S P A E D S S P G I S L S S S R G P G R
AAGTGCCGCTGAGGAGCTTGACGAGGAAATACCCGTATCTTCCCACTACTTTGCAAGTAAAACCAGAAATGAAAGGAAGA 4080
1310 S A A E E L D E E I P V S S H Y F A S K T R N E R K
GGAAAAAGATGCCAGCCTCCCAAAGGTCTAAGAGGAGAAAAACTGCTTCCAGTGGTTCCAAGGCAAAGGGGGGGTCTGCC 4160
1336 R K K M P A S Q R S K R R K T A S S G S K A K G G S A
ACATGTAGAAAGATATCTTCCAAAACGAAATCCTCCAGCATCATTGGATCCAGTTCAGCCTCACATACTTCTCAAGCGAC 4240
1363 T C R K I S S K T K S S S I I G S S S A S H T S Q A T
ATCAGGAGCCAATAGCAAATTGGGGATTATGGCTCCACCGAAGCCTATAAATAGACCGTTTCTTAAGCCTTCATATGCAT 4320
1390 S G A N S K L G I M A P P K P I N R P F L K P S Y A
TCTCATAAcaaccgaatctcaatgtacatagaccctctttcttgtttgtcagcatctgaccatctgtgactataaagctg 4400
1416 F S
ttattcttgttataccaaaaaaaaaaaaaaaaaaaaa 4437

FIG. 2D

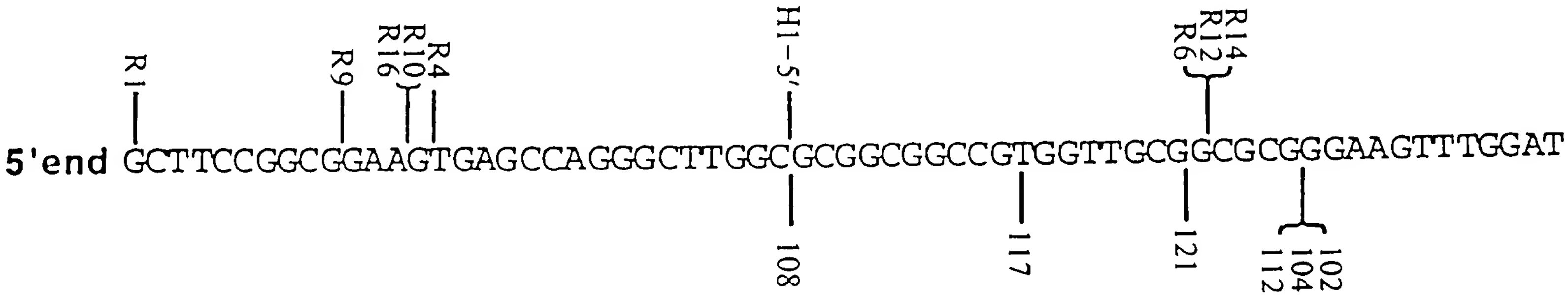
5'end GCTTCCGGCGGAAGTGAGCCAGGGCTTGGCGCGGCGGCCGTGGTTGCGGCGCGGAAGTTTGGAT
R1
R9
R10
R16
R4
H1-5'
R14
R12
R6
108
117
121
102
104
112

FIG. 3

```
                            *                 I                             Ia
649  FPHTKEMMKIFHKKFGLHNFRTNQLEAINAALLGEDCFILMPTGGGKSLCYQLPACV----SPGVTVVISPLRSLIVDQV BLM
 74  FPWSGKVKDILQNVFKLEKFRPLQLETINVTMAGKEVFLVMPTGGGKSLCYQLPALC----SDGFTLVICPLISLMEDQL REQL
659  YPWSDEVLYRLHEVFKLPGFRPNQLEAVNATLQGKDVFVLMPTGGCKSLCYQLPAVVKSGKTHGTTIVISPLISLMQDQV SGS1
 16  ---------VLQETFGYQQFRPGQEEIIDTVLSGRDCLVVMPTGGGKSLCYQIPALL----LNGLTVVVSPLISLMKDQV recQ

                                                                         II
725  QKLTSLDIPATYLTGDKTDSEATNIYLQLSKKDPIIKLLYVTPEKICASNRLISTLENLYERKLLARFVIDEAHCVSQWG BLM
150  MVLKQLGISATMLNASSSKEHVKWVHDEMVNKNSELKLIYVTPEKIAKSKMFMSRLEKAYEARRFTRIAVDEVHCCSQWQ REQL
739  EHLLNKNIKASMFSSRGTAEQRRQTFNLFIN--GLLDLVYISPEMISASEQCKRAISRLYADGKLARIVVDEAHCVSNWG SGS1
 83  DQLQANGVAAACLNSTQTREQQLEVMT--GCRTGQIRLLYIAPERL----MLDNFLEHL-AHWNPVLLAVDEAHCISQWG recQ

                                   III    *
805  HDFRQDYKRMNMLRQKFPSVPVMALTATANPRVQKDILTQLKILRPQVFSMSFNRHNLKYYVLPKKPKKVA---FDCLEW BLM
230  HDFRPDYKALGILKRQFPNASLIGLTATATNHVLTDAQKILCIEKCFTFTASFNRPNL-YYEVRQKPSNTEDFIEDIVKL REQL
817  HDFRPDYKELKFFKREYPDIPMIALTATASEQVRMDIIHNLELKEPVFLKQSFNRTNL-YYEVNKKTKNT---IFEICDA SGS1
157  HDFRPEYAALGQLRQRFPTLPFMALTATADDTTRQDIVRLLGLNDPLIQISSEDRPNIRY-MLMEKFKPLDQLM----RY recQ

                    IV                                                  V
882  IRKHHPYDSGIIYCLSRRECDTMADTLQRDGLAALAYHAGLSDSARDEVQQKWINQDGCQVICATIAFGMGIDKPDVRFV BLM
309  INGRYKGQSGIIYCFSQKDSEQVTVSLQNLGIHAGAYHANLEPEDKTTVHRKWSANE-IQVVVATVAFGMGIDKPDVRFV REQL
893  VKSRFKNQTGIIYCHSKKSCEQTSAQMQRNGIKCAYYHAGMEPDERLSVQKAWQADE-IQVICATVAFGMGIDKPDVRFV SGS1
233  VQEQ-RGKSGIIYCNSRAKVEDTAAALQSKGISAAAYHAGLENNVRADVQEKFQRDD-LQIVVATVAFGMGINKPNVRFV recQ

                    VI
962  IHASLPKSVEGYYQESGRAGRDGEISHCLLFYTYHDVTRLKRLIMMEKDGNHHTRETHFNNLYSMVHYCENITECRRIQL BLM
388  IHHSMSKSMENYYQESGRAGRDDMKADCILYYGFGDIFRISSMVVMENVGQQ--------KLYEMVSYCQNISKSRRVLM REQL
972  YHFTVPRTLEGYYQETGRAGRDGNYSYCITYFSFRDIRTMQTMIQKDKNLDRENKEKHLNKLQQVMAYCDNVTDCRRKLV SGS1
311  VHFDIPRNIESYYQETGRAGRDGLPAEAMLFYDPADMAWLRRCLEEKPQGQLQDIERH--KLNAMGAFAEAQT-CRRLVL recQ
```

FIG. 4

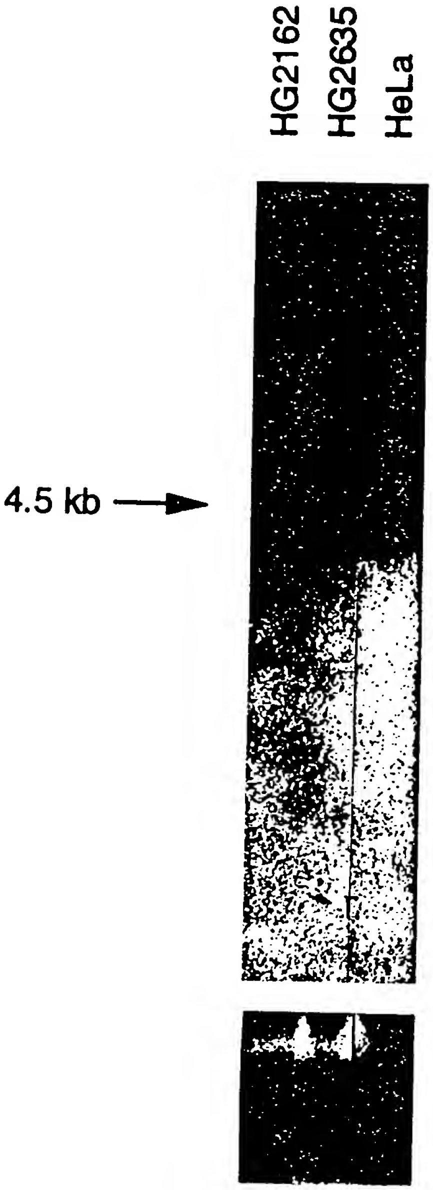
HG2162
HG2635
HeLa
4.5 kb

FIG. 5A

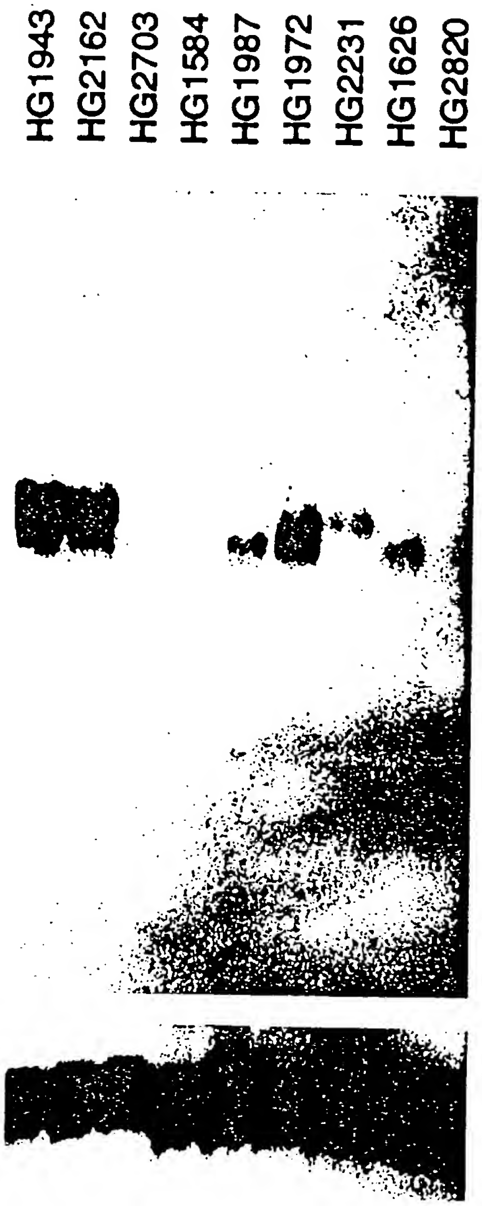
HG1943
HG2162
HG2703
HG1584
HG1987
HG1972
HG2231
HG1626
HG2820

FIG. 5B

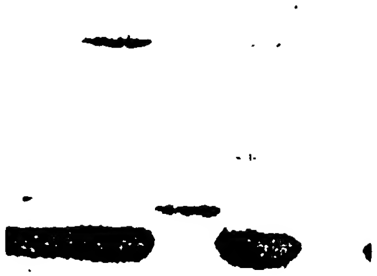

FIG. 6A

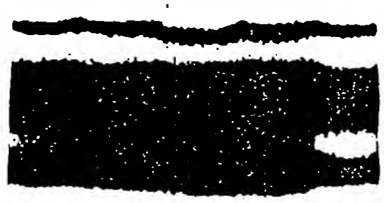

FIG. 6B

FIG. 6C

FIG. 6D

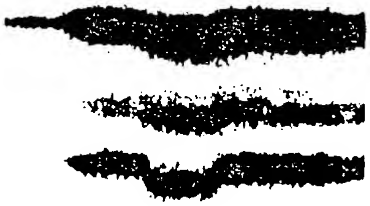

FIG. 6E